Family Health History Tool Meeting 2016 Meeting Summary

The National Human Genome Research Institute (NHGRI)

By Bob Wildin, Chief of the Genomic Healthcare Branch

On June 14th and 15th, NHGRI convened a two-day conference at NIH exploring the state of the electronic family health history tool field. These "tools," as I will call them here, are computer programs that help collect and organize a family's health history. Such information can be used to assess the risks of conditions influenced by heredity and to inform health planning and healthcare decisions made by families and their healthcare providers.

The overarching goal was to prepare the tools field to improve personal health by responding effectively to rapid changes in Family Health History (FHH) data uses, Health Information Technology (HIT) capabilities, and research opportunities. This conference aimed to identify and share successful approaches to using family health history tools, and to identify unresolved issues and potential solutions that may be addressed by policy, research, and/or collaborative efforts.

The meeting was attended by about 70 participants representing developers and vendors of 14 tools, patient advocates, subject matter experts, researchers, information technology specialists, one electronic health record system (EHR) vendor, and representatives from stakeholder Federal and state agencies. About 55 participants attended in person, with the rest connecting via teleconferencing with screen sharing.

In the introductory session, the meeting was brought to order by Laura Lyman Rodriguez, Ph.D., Director, Division of Policy, Communications, and Education at NHGRI. Bob Wildin, M.D., Chief of the Genomic Healthcare Branch under Dr. Rodriguez, and organizer of this gathering, outlined the meeting's structure and proposed a framework for thought and discussion. Dr. Greg Feero of Main General Hospital, and previously at NHGRI and the National Cancer Institute (NCI), reviewed the history of My Family Health Portrait, a.k.a. The Surgeon General's Family History Tool, which was one of the first such tools developed. It was created to help raise public awareness of the importance of this free information for optimizing risk detection and management. Dr. Muin Khoury, of the Centers for Disease Control (CDC), discussed the importance of FHH in public health and disease prevention, and Megan Doerr, Certified Genetic Counselor, described the efforts of the Global Alliance for Genomic Health (GA4GH) working group focused on FHH, including development of the first family history tool inventory in 2015 that was a key resource for developing the meeting's structure and content.

Tool developers and vendors (Table 1) presented their tools to the audience, followed by live, interactive demonstrations in smaller groups. Attendees were able to experience multiple tools, ask questions, and understand their unique features and strengths. The tools included stand-alone applications with and without Health-Level 7 (HL7) standards compliance, applications designed or adapted for electronic health record (EHR) integration, ones intended for lay and professional users,

including low-literacy and non-English users, and tools that include various levels of Clinical Decision Support (CDS). CDS was most often implemented in tools targeted for cancer risk assessment. Tools were often developed to facilitate an efficient FHH collection and use workflow in clinical settings. The traditional staff-intensive workflow is often cited as a reason for low compliance with professional guidelines for the use of FHH in clinical care. Most tools were designed specifically to support the FHH practice gap in healthcare settings. One intriguing system is a Personal Health Record (PHR) that allows individuals to consent to reveal their PHR data for specific relatives, creating a *de facto* family health history record for them.

In between tool demonstration sessions, invited experts (Table 2) gave presentations illuminating various facets of the FHH field: recent advances, challenges, and opportunities. Topics included HIT interoperability and HL7 standards, quality of and discrepancies in data collected, added value from using health histories and genomic variant information together, modern family structure variation, expanded uses including public health, adapting for low literacy users, and the ethics and regulatory landscape for sharing health and other sensitive data among family members. Vigorous and informative discussions followed each speaker session.

Use and integration of professional guidelines in tool-linked computer algorithms to calculate disease risk estimates for common diseases, i.e. CDS, and how to link tools with prevalent electronic health record (EHR) systems, were the topics of expert panel discussions with equally energized audience participation. Combined, these systems may greatly amplify the capacity of health providers and health systems to implement recommended FHH risk detection guidelines and the genetic testing and counseling indicated after detection of increased risk. They may also facilitate direct FHH data entry by the patient and raise their awareness of risk, easing the burden on providers with limited visit times. One potential drawback is that the quality of the data so collected may be quite variable.

Breakout groups engaging participants in deeper discussion of their favorite topic topped off the meeting. Groups were charged with reporting observations and recommendations from their group to the rest of the attendees. The Data Standards group recommended educational campaigns on standards, promoting getting started with HL7 Fast Healthcare Interoperability Resource (FHIR), and using recognized workflows to enumerate necessary standards uses.

The EHR group asserted that personalized medicine requires FHH as a foundation and proposed that coverage for genetic tests be dependent on a minimum standard FHH collection prior to testing. As an additional barrier to obtaining indicated testing, some opposed this. They pointed to the power of EHR buyers and end-users (providers, payers, and employers) to insist on acceptable FHH functionality, and discussed the level of FHH function required within EHRs as compared to using separate family health history tool systems tightly linked to EHRs for collection, presentation, and CDS.

The CDS group distilled their recommendations to one: Medical societies and government agencies publishing clinical practice guidelines should be certain those guidelines can be converted to algorithms that can be implemented in CDS software, i.e., contain no ambiguous language and be machine readable.

The Policy group celebrated past milestones such as privacy legislation (GINA, HIPAA), improved healthcare access, and HIT developments including patient portals. They touched on an extended list of policy issues and opportunities that directly or indirectly limit optimal use of FHH information for

healthcare, including the need for quality metrics and possible incentives for use. Priority areas included reimbursement for genetics experts for supporting the quality FHH process and its outcomes, engaging the President's Precision Medicine Initiative to advance the use of FHH for research and healthcare, highlight the privacy conversation, and explore the public health implications of FHH. Developing privacy use-cases that could be used by the public and regulators to explore the ethical limits of opportunities for intra-family health data sharing was also identified as key to supporting forward progress.

The Research funding opportunities group suggested engaging PCORI more in this field, and noted the lack of studies of family health history tools' impact on healthcare utilization and short- and long-term cost. It emphasized the need for validation of effectiveness of the individual FHH tools on the market, by the tool's developer at minimum. This group also listed challenges to patient engagement and the need to publish existing data. Workflows that emphasize FHH early in the patient appointment process show promise. Additional consideration for removing barriers in health disparity and low literacy populations is needed. The group's participants observed that additional technological approaches to FHH collection could be explored or expanded, including remote scribes and automated telephone interactive voice response systems. They also commented on the shrinkage and narrow targeting of public funding for public health genomics, including FHH use. Only three states are using limited public health approaches to cancer surveillance and or engagement.

Conclusions and Next Steps

Attendees and presenters commented on the rapid pace of change in the field, with some tools coming into regular clinical and commercial use. The sophistication and value of tools has increased considerably in the past few years. While the tools, as a whole, can clearly facilitate implementation of clinically-appropriate collection and use of FHH, a large gap remains between the current state and optimal adoption of family history risk-based care. Substantial focus was brought during the meeting on several key areas amenable to current action, as outlined above. Future progress depends on continued work on the tools themselves, but as importantly, on collaborative efforts to pave the way for all tool developers and vendors. In particular, the data-interoperability standards problem has solutions available and more on the horizon. Incorporation of CDS is challenging due to imprecision in the language of guidelines on which they are patterned, but shows great promise for creating efficient workflows and greater uptake of evidence-based practice recommendations.

The meeting organizers, speakers, and tool presenters will work together to publish more detailed papers focusing on the status of Family History usage and assistive technologies, on the range of tools available for different settings, and on the important policy issues affecting the field. Follow-on conversations are planned for the now-monthly Fed/Non-Fed Family History Group conference calls, with the intent of further fostering collaboration, especially between stakeholders from different sectors. Further engagement in the policy opportunities highlighted here will be sought.

The complete agenda for the meeting, presenters' slides, and tool descriptions are available on the meeting website: https://www.genome.gov/27565264/the-nih-family-health-history-tool-conference-2016/.

Table 1. Family Health History Tools and Tool Presenters

Family Health History Tool	Tool Presenter/Demonstrator	Affiliation (alphabetical)
VICKY	Catherine Wang, PhD	Boston University
CancerIQ	Haibo Lu, MBA	Cancer IQ, Inc
	Feyi Olopade Ayodele, MBA	Cancer IQ, Inc.
PROBAND	Jeffrey Miller, BS	Children's Hospital of Philadelphia
CRA Health (formerly Hughes RiskApps)	Kevin Hughes, MD	CRA Health, LLC
MeTree	Lori A. Orlando, MD, MHS	Duke University
Hughes Risk Apps: Tablet	Kevin Hughes, MD	Massachusetts General Hospital
Hereditary Cancer Quiz	Karli Slocum, BS	Myriad Genetic Laboratories, Inc.
Myriad Family History Tool	Randall Adams	Myriad Genetics
	Jeremy Bennett, MS, PMST	Myriad Genetics
	Leigh Baumgart MS, PhD	NantHealth, LLC
Health Heritage	William A. Knaus, MD	NantHealth, LLC
My Family Health Portrait	Bob Wildin, MD	National Human Genome Research Institute
	Donna Messersmith, PhD	National Human Genome Research Institute
CancerGene Connect	Richard Burghardt	OMMDOM, Inc.
	Bentley Davis	OMMDOM, Inc.
	Megan Frone, MS, CGC	OMMDOM, Inc.
Progeny Family History Questionnaire (FHQ)	Michael Brammer	Progeny Genetics, LLC
Family Healthware	Raymond Solone, MS	Sanitas, Inc.
Zibdy Health	Hirdey Bhathal, MS, PhD	Zibdy Health

Table 2. Invited Speakers and Panel Members

Speaker (alphabetical)	Affiliation
Robin L. Bennett, MS, CGC	University of Washington Medical Center, Division of Medical Genetics
Hirdey Bhathal, MS, PhD	Zibdy Health
Michael Brammer	Progeny Genetics, LLC
Megan Doerr, MS	Sage Bionetworks
David Dubin	AliveAndKick'n
Gregory Feero, MD, PhD	Maine Dartmouth Family Medicine Residency
Marissa Gordon-Nguyen, MPH, JD	Department of Health and Human Services, Office for Civil Rights
Kevin Hughes, MD	Massachusetts General Hospital
Muin J. Khoury, MD, PhD	Director, CDC Office of Public Health Genomics
Laura Koehly, PhD	National Human Genome Research Institute
Howard Levy, MD, PhD	Johns Hopkins University School of Medicine
Lori Orlando, MD	Duke University School of Medicine
Laura L. Rodriguez, PhD	National Human Genome Research Institute
Brian Shirts, MD, PhD	University of Washington
Catharine Wang, PhD	Boston University School of Public Health
Thomas Weber, MD	SUNY Institute for Genomic Health & Department of Surgery

Bob Wildin, MD	National Human Genome Research Institute
Grant Wood	Intermountain Health Care